

## Clinical re-biopsy of segmental gains, the primary source of preimplantation genetic testing false positives

Journal of Assisted Reproduction and Genetics

**Supplementary table 1 VeriSeq PGT-A SA detection limits**

Translocation breakpoint	Size (Mb) <sup>a</sup>	Gain (G)/Loss (L)	Observed shift
17q25.3	2.0	L	30%
21q22.3	3.6	G	50%
21q22.3	3.6	L	60%
16p13.3	3.8	L	30%
21q21.3	3.8	G	50%
3p26.2	4.0	L	60%
3p26.2	4.0	G	40%
7p22.3	4.0	G	40%
4q28.3 (interstitial)	4.7	L	60%
1q42.13-q42.3 (interstitial)	6.9	G	90%
22q13.31	9.3	L	80%
12p13.1	8.7	G	70%
17p13.2	9.1	L	80%
19p13.2	9.6	L	110%
5p15.31	10.4	G	70%
4p16.1	11.2	G	90%
3p25.1	16.1	L	100%
9q33.2	16.0	G	90%
Xq26.3	17.9	G	80%
Xq26.3	17.9	L	100%

<sup>a</sup> as determined by array CGH enriched with telomeric probes for the detection of small unbalanced translocations.

**Supplementary table 2 VeriSeq PGT-A mosaic SA detection limits**

Aneuploidy	Size (Mb)	Observed shift at 100% titration	Detection limits	Notes on titrations
-6q26-q27	9.0	100%	≥50%	10-20% undetectable, 30% difficult to distinguish from background
+5p15.33-p15.31	10.4	75%	≥50%	10% undetectable, 20-30% difficult to distinguish from background
-12q24.31-q24.33	11.0	95%	≥30%	10% undetectable, 20% difficult to distinguish from background
-1p36.33-p36.21	16.0	95%	≥30%	10-20% undetectable
-3p26.3-p25.1	16.1	100%	≥20%	10% undetectable
+22q12.2-q13.33	21	95%	≥30%	10% undetectable, 20% difficult to distinguish from background
-13p13-q14.11	24	100%	≥30%	10% undetectable, 20% difficult to distinguish from background
+5q33.2-q35.3	25	95%	≥20%	10% difficult to distinguish from background
+20p13-p11.1	28	105%	≥10%	All titrations clearly visible.
+4p16.3-p14	41	105%	≥20%	10% undetectable

**Supplementary table 3 PGT-A testing outcomes**

Group	Description	Count	Percentage	Age at OPU <sup>a</sup>	PGT-M <sup>d</sup>
ABNn	Abnormal, numeric chromosome(s)	4343	29.5%	38.1±3.8 <sup>b</sup>	436 (10.0%)
ABNs	Abnormal, SA(s)	468	3.2%	35.2±3.8	67 (14.3%)
ABNb	Abnormal, numeric chromosome(s) + full SA(s)	284	1.9%	37.8±3.9 <sup>b</sup>	38 (13.4%)
A	NAD, clear PGT-A profiles	4690	31.9%	35.1±3.8	688 (14.7%)
B1	NAD, noisy PGT-A profiles/mosaicism <20%	1467	10.0%	35.2±3.8	217 (14.7%)
B2	Mosaic, SA(s) 20-80%	1209	8.2%	35.1±3.9	165 (13.6%)
B3	Mosaic, numeric chromosome(s) 20-40%	896	6.1%	35.1±3.7	123 (13.7%)
B3 +SA	Mosaic, numeric chromosome(s) 20-40% + mosaic SA(s) 20-40%	325	2.2%	35.2±3.7	41 (12.6%)
C	Mosaic, numeric chromosome(s) >40-<80%	473	3.2%	35.9±3.9 <sup>b</sup>	74 (15.6%)
C +SA	Mosaic, numeric chromosome(s) >40-<80% + mosaic SA(s) 20-<80%	103	0.7%	35.7±4.3	7 (6.8%)
UNK	Test failed	447	3.0%	35.7±4.2 <sup>c</sup>	61 (13.6%)
Total		14705	100% <sup>e</sup>	36.1±4.1	1917 (13.0%)

<sup>a</sup>mean ± SD of female age at OPU (oocyte pick up), <sup>b</sup>P < 0.0001 (t-test) vs group A (NAD), <sup>c</sup>P < 0.05 vs group A (t-test), <sup>d</sup>proportion of samples that underwent parallel PGT-M analysis, <sup>e</sup>total adds to 99.9% due to rounding errors.

**Supplementary table 4 Pregnancy outcomes of mosaic subcategories**

Group	B2 low	B2 high	B2 single	B2 multiple	B2 gain	B2 loss	B3 single	B3 multiple
Description	20-40% SA	41-79% SA	20-79% one SA	20-79% >1 SA	20-79% SA duplication	20-79% SA deletion	20-40% one numeric	20-40% >1 numeric
Age	36.1±4.0	37.1±3.3	35.9±3.9	37.0±3.7	36.1±3.7	35.8±4.1	36.0±3.7	35.5±3.8
Day 6	36.8%	48.0%	38.8%	35.1%	38.6 %	37.4 %	42.6%	46.8%
Grade 1	11.0% (20)	16.0% (4)	12.4% (21)	8.1% (3)	14.3% (10)	9.8% (12)	6.1% (7)	8.7% (11)
Grade 2	23.6% (43)	16.0% (4)	22.4% (38)	24.3% (9)	27.1% (19)	21.1% (26)	21.7% (25)	26.2% (33)
Grade 3	53.8% (98)	52.0% (13)	51.2% (87)	64.9% (24)	51.4% (36)	53.7% (66)	58.3% (67)	47.6% (60)
Grade 4	11.5% (21)	16.0% (4)	14.1% (24)	2.7% (1)	7.1% (5)	15.4% (19)	13.9% (16)	17.5% (22)
Transferred	182	25	170	37	70	123	115	126
+ve bHCG	63.2% (115)	76.0% (19)	66.5% (113)	56.8% (21)	72.9% (51)	64.2% (79)	60.0% (69)	52.4% (66)
SCMC	13.0% (15/115)	10.5% (2/19)	12.4% (14/113)	14.3% (3/21)	13.7% (7/51)	12.7% (2/79)	21.7% (15/69)	18.2% (12/66)
+ve FHP	55.0% (100)	68.0% (17)	58.2% (99)	48.7% (18)	62.9% (44)	56.1% (69)	47.0% (54)	42.9% (54)
CM	5.0% (5/100)	0% (0/17)	4.0% (4/99)	5.7% (1/18)	6.8% (3/44)	2.9% (2/69)	7.4% (4/54)	3.7% (2/54)
LB rate	52.2% (95/182)	68.0% (17/25)	55.9% (95/170)	46.0% (17/37)	58.6% (41/70)	54.5% (67/123)	43.5% (50/115)	41.3% (52/126)
LB P <sup>a</sup>	0.14		0.27		0.58		0.73	

bHCG; positive bHCG, FHP; foetal heart pregnancy, SCMC; sub-clinical miscarriage (bHCG-FHP), CM; clinical miscarriage (FHP-LB), LB; livebirth, <sup>a</sup> P value (Chi-Square).

**Supplementary table 5 Matched mosaic pregnancy outcomes**

Group	A	B1	B2	B3	C
Description	NAD (high quality PGT-A profiles)	Noisy/mosaicism <20%	20-79% segmental mosaicism	20-40% numeric chr +/- segmental	41-79% numeric chr +/- segmental
Age <sup>a</sup>	36.3±3.7	36.3±3.7	36.2±4.2	36.1±4.0	37.0±3.1
Day 6 Biopsy (%)	60.8	61.9	61.2	62.5	61.5
Grade 1	4.0% (16)	4.0% (5)	4.5% (3)	4.2% (4)	3.8% (1)
Grade 2	4.0% (16)	4.0% (5)	4.5% (3)	4.2% (4)	3.8% (1)
Grade 3	53.3% (212)	54.0% (68)	53.7% (36)	54.2% (52)	53.8% (14)
Grade 4	38.7% (154)	38.1% (48)	37.3% (25)	37.5% (36)	38.5% (10)
Transferred	398	126	67	96	26
PGT-M <sup>b</sup>	11.1% (44)	14.3% (18)	11.9% (8)	12.5% (12)	7.7% (2)
+ve bHCG	65.6% (261)	65.9% (83)	67.2% (45)	53.1% (51)	38.5% (10)
SCMC	11.9% (31/261)	19.3% (16/83)	15.6% (7/45)	15.7% (8/51)	40.0% (4/10)
SCMC P <0.1	A vs B1: 0.088, A vs C: 0.009 <sup>c</sup> , B2 vs C: 0.052, B3 vs C: 0.083				
+ve FHP	57.8% (230)	53.2% (67)	56.7% (38)	44.8% (43)	23.1% (6)
CM	7.0% (16/230)	1.5% (1/67)	0% (0/38)	7.0% (3/43)	16.7% (1/6)
CM P <0.1	A vs B1: 0.091, A vs B2: 0.094, B1 vs C: 0.03 <sup>c</sup> , B2 vs B3: 0.099				
LB rate	53.8% (214/398)	52.4% (66/126)	56.7% (38/67)	41.7% (40/96)	19.2% (5/26)
LB P vs A	-	0.79	0.65	0.033 <sup>c</sup>	<0.001 <sup>c</sup>
Other LB P <0.1	B1 vs C: 0.002 <sup>c</sup> , B2 vs B3: 0.059, B2 vs C: 0.001 <sup>c</sup> , B3 vs C: 0.036 <sup>c</sup>				

bHCG; positive bHCG, FHP; foetal heart pregnancy, SCMC; sub-clinical miscarriage (bHCG-FHP), CM; clinical miscarriage (FHP-LB), LB; livebirth, <sup>a</sup> all age comparisons were not significant (t-test), <sup>b</sup> proportion of samples that underwent parallel PGT-M analysis, <sup>c</sup>P < 0.05 (Chi-Square or Fisher's exact).

**Supplementary table 6 Full details of initial cohort of SA blastocysts thawed for clinical rebiopsy**

Sample	Fate	Grade	Day	SA type <sup>a</sup>	Original result (% = shifts other than 100%) <sup>b</sup>	Size (Mb) <sup>c</sup>	Re-biopsy result <sup>d</sup>	New status <sup>e</sup>
1	Re-biopsy	HB 2-3	5	G	+7q34-q36.3	20	NAD	Mosaic
2	Discard	HB 2-2	6	L complex	-17q23.2-q25.3, +17q22-q23.2 (70%)	63	na	na
3	Discard	HB 2-3	6	L complex	-11q21-q25, +11p15.5-q21 (55%)	37	na	na
4	Discard	XB 2-2	5	B complex	+11q21-q24.2, -11q24.2-q25, +11p15.5-q21 (30%)	9, 24	na	na
5	Discard	HB 2-3	5	L	-15q14-q26.3	67	na	na
6	Discard	HB 2-3	6	L	-20p13-p11.22	22	na	na
7	Re-biopsy	HB 2-2	5	B	+16p13.3-p12.3 (165%), -16p12.3-q24.3	18, 71	No call	na
8*	Re-biopsy	HB 1-2	5	L complex	-3p26.1-p25.3, -3p26.3-p26.1 (60%), +3q25-1-q29 (30%)	6	-3p26.1-p25.3, -3p26.3-p26.1 (60%)	ABN
9*	Re-biopsy	FHB 2-2	6	L	-5p15.33-p13.3	30	-5p15.33-p15.1, -5p15.1-q35.3 (70%)	ABN
10	Re-biopsy	HB 2-2	5	G	+Xq27.1-q28	15	No call	na
11*	Re-biopsy	HB 2-3	6	G complex	+3q22.3-q26.32 (90%), +3q26.33-q29 (125%)	60	+3q12.3-q29 (135%)	ABN
12*	Re-biopsy	HB 2-2	5	G	+3q11.2-q29	180	-3p14.3-q29 (20%)	Mosaic
13*	Re-biopsy	HB 2-2	5	L	-3q25.1-q29, -3p26.3-q25.1 (30%)	48	-3q25.1-q29, -3p26.3-q25.1 (60%)	ABN
14	Re-biopsy	FHB 2-2	6	L	-4p16.3-p13	44	-4p16.3-p13	ABN
15*	Re-biopsy	HB 1-2	5	L complex	-3q23-q29, -3p26.3-q23 (35%)	55	-3q23-q29	ABN
16	Re-biopsy	HB 2-2	6	G complex	+11q24.2-q25 (230%), -11p15.5-q24.2 (30%)	9	NAD	Mosaic
17	Re-biopsy	FHB 2-1	6	L	-6q27-q27	5	-6q27-q27	ABN
18*	Re-biopsy	HB 2-3	6	L complex	-16p13.3-p12.3, -16p12.3-q24.3 (50%)	18	-16p13.3-p12.3	ABN
19	Discard	HB 1-2	6	L complex	-16q12.1-q23.3, -16p13.3-q11.2 (70%), -16q24.1-q24.3 (65%)	38	na	na
20*	Re-biopsy	HB 1-1	5	L complex	-5q12.1q35.3, -5p15.33-q11.2 (35%)	120	-5q12.1q35.3, +5p15.33-q11.2 (55%)	ABN
21*	Re-biopsy	HB 1-2	6	L complex	-21q21.2-q22.3, +21p13-q21.2 (55%)	22	-21q21.2-q22.3, -21p13-q21.2 (60%)	ABN
22	Re-biopsy	HB 1-1	5	G	+11q23.2-q25	20	NAD	Mosaic
23*	Re-biopsy	HB 2-2	5	G	+5q11.2-q35.3 (130%)	132	+5q11.2-q35.3 (25%)	Mosaic
24	Re-biopsy	HB 1-2	5	G	+8q11.21-q24.3 (1800%)	95	NAD	Mosaic
25	Discard	HB 2-2	6	G	-21q22.12-q22.3	11	na	na
26	Discard	HB 2-3	6	L	-2q22.1-q37.3	111	na	na
27*#	Re-biopsy	HB 1-2	5	L complex	-11q23.3-q25 (85%), -11p15.5-q23.3 (55%)	17	-11 (70%)	Mosaic

28	Discard	HB 2-2	5	B	+9q31.1-q31.3, -9q33.3-q34.3	8, 14	na	na
29	Re-biopsy	HB 2-2	5	L	-10q11.22-q26.3	86	-10q11.22-q26.3	ABN
30	Re-biopsy	HB 2-3	5	G	+1p36.33-q12	133	NAD	Mosaic
31*	Re-biopsy	FHB 2-2	6	L complex	-10q26.11-q26.3, -10p15.3-q25.3 (55%)	15	-10q26.11-q26.3, -10p15.3-q25.3 (85%)	ABN
32*	Re-biopsy	HB 2-2	6	B	+6p25.3-q23.1, -6q23.1-q27	130, 44	-6q23.1-q27	ABN
33*	Re-biopsy	HB 2-2	6	L complex	-9p24.3-p21.1, -9p13.3-q34.3 (25%)	30	-9p24.3-p21.1	ABN
34	Re-biopsy	FHB 2-2	6	G	6q16.1-q27 (140%)	75	NAD	Mosaic
35	Re-biopsy	HB 2-2	5	L complex	-7q31.1-q36.3, -7p22.3-q31.2 (35%)	45	-7q31.1-q36.3, -7p22.3-q31.2 (35%)	ABN
36*	Re-biopsy	HB 2-2	5	G complex	+18q22.2-q23 (575%) +18p11.32-q12.1 (25%)	11	+18q22.2-q23 (250%), -18p11.32-q22.2,	ABN
37	Re-biopsy	HB 1-2	6	L complex	-8p23.3-p11.23, -8p11.22-q24.3 (65%)	37	-8p23.3-p11.23, -8p11.22-q24.3 (65%)	ABN
38*	Re-biopsy	HB 2-2	5	B	-5q34-q35.3, +15q21.1-q26.1	13, 33	-5q34-q35.3, +15q21.1-q26.1, -5q11.2-q34 (50%)	ABN
39*	Re-biopsy	HB 1-2	5	G complex	+20q11.21-q13.33, +20p13-p11.21 (70%)	33	+20	ABN
40	Re-biopsy	HB 2-2	6	G	+2q34-q37.3 (130%)	33	NAD	Mosaic
41*	Re-biopsy	HB 2-2	6	L complex	-Xp21.3-Xp21.1, -Xq21.1-q21.33, -Xp22.33-p22.11 (70%), -Xp11.4-q13.3 (70%), -Xq22.1-q28 (70%)	11, 20	XO	ABN
42*	Re-biopsy	HB 1-3	6	L	-1p36.33-p33	50	-1p36.33-p33, -1p32.3-q44 (60%)	ABN
43	Discard	HB 2-3	5	L complex	-5q11.2-q35.3, -5p15.33-p12 (25%)	110	na	na
44*	Re-biopsy	HB 2-2	5	G	+9p24.3-p11.2 (150%)	41	+9p24.3-q21.11 (25%)	Mosaic
45*	Re-biopsy	HB 1-2	6	L	-7q22.1-q36.3	55	-7q22.1-q36.3 (50%)	Mosaic
46	Discard	HB 2-1	6	L	-1q12-q44	102	na	na
47	Re-biopsy	HB 2-2	5	G complex	+8p23.1-p21.2 (150%), +8p21.2-q24.3 (85%)	134	+8p23.1-p21.2 (150%), +8p21.2-q24.3 (85%)	ABN
48	Discard	HB 2-3	6	L complex	-1q31.1-q44, +1p36.33-q25.3 (30%)	63	na	na
49	Discard	HB 2-2	5	L complex	-12q13.13-q24.33, -12p13.33-q13.12 (25%)	70	na	na
50#	Re-biopsy	HB 2-2	5	L	-2q33.1-q37.3 (90%)	34	NAD	Mosaic
51	Re-biopsy	HB 1-2	5	L complex	-10q23.31-q26.3, -10p15.3-q23.31 (45%)	29	No call	na
52	Discard	HB 2-2	5	L	-6q25.3-q27	14	na	na
53*	Re-biopsy	FHB 2-3	6	G	+2q33.3-q37.3	34	+2 (35%)	Mosaic
54	Re-biopsy	HB 2-2	5	G	+4p16.3-p16.1	8	NAD	Mosaic
55*	Re-biopsy	HB 1-2	6	L complex	-8q22.3-q24.3, -8p23.3-q22.3 (35%)	41	-8p23.3-q22.3 (45%), +8q22.3-q24.23 (25%)	Mosaic
56	Re-biopsy	HB 2-3	6	L complex	-13q21.31-q34, -13p13-q21.2 (25%)	51	-13q21.31-q34, -13p13-q21.2 (25%)	ABN

57	Re-biopsy	HB 2-2	5	B	-8p23.3-p23.1, +8p22-p12	12	-8p23.3-p23.1, +8p22-p12	ABN
58*	Re-biopsy	HB 2-2	5	G complex	+10p15.3-p11.22, +15p13-q23	32, 72	+10p15.3-q23.1, +15p13-q25.2 (75%)	ABN
59#	Re-biopsy	HB 1-1	5	L	-1q43-q44 (80%)	10	NAD	Mosaic
60*	Re-biopsy	HB 2-2	6	L	-16q11.2-q24.3	51	-16q12.1-q24.3 (80%), -16p13.3-q11.2 (30%)	ABN
61*	Re-biopsy	HB 1-2	5	G	+9p21.1-q34.3 (270%)	106	+9p23-q12 (80%)	ABN
62*	Re-biopsy	HB 2-2	6	L complex	-19p13.3-p13.12, +19p13.12-q13.43 (40%)	13	-19p13.3-p13.12, -19p13.12-q13.43 (55%)	ABN
63*	Re-biopsy	HB 2-2	5	G complex	+22p13-q12.3, +22q13.1-q13.33 (60%)	37	+22	ABN
64*	Re-biopsy	HB 2-2	5	B	-10q26.13-q26.3	10	Chaotic chromosomal abnormalities (not involving chromosome 10)	ABN
65	Discard	HB 2-3	6	L	-1p36.33-p35.2	31	na	na
66	Discard	HB 2-3	6	L complex	-10q25.2-q26.3, -10p15.3-q25.1 (55%)	21	na	na
67#	Re-biopsy	HB 2-3	5	L complex	-10q21.1-q26.3 (90%), -13p13-q14.11 (85%)	81, 47	NAD	Mosaic
68*	Re-biopsy	HB 3-2	6	B	-3q13.33-q34, +3p13-q13.33 (85%)	77	-3q13.33-q34	ABN
69*	Re-biopsy	HB 2-2	5	L	-7p22.3-p15.3	24	-7p22.3-p12.3, -7p12-q36.3 (80%)	ABN
70*	Re-biopsy	HB 2-2	5	B	+Xq27.2-q28 (300%), -Xp22.33-q27.1	15, 140	+Xq27.2-q28 (45%), -Xp22.3-q27.1	ABN
71*	Re-biopsy	HB 1-2	5	L complex	-5p15.33-p15.1, -5p14.3-q23.2 (40%), -5q23.2-q35.3 (75%)	16	+5 (25%)	Mosaic
72	Discard	HB 2-2	5	G complex	+4q28.3-q35.2 (130%), -4p16.3-q28.2 (20%)	57	na	na
73	Re-biopsy	HB 2-2	6	L	-13q31.1-q34	33	-13q31.1-q34	ABN
74	Discard	HB 2-2	5	L complex	-6p25.3-p21.31, -6p21.2-q27 (25%)	36	na	na
75*	Re-biopsy	HB 2-2	6	B	+18p11.32-p11.31, -18p11.31-q23	4	-18p11.32-p11.31	ABN
76	Re-biopsy	HB 1-2	5	B	-9p24.3-q13, +9q21.11-q34.3 (80%)	63, 71	NAD	Mosaic
77*	Re-biopsy	HB 2-2	5	L complex	-2q31.1-q37.3, 2p25.3-q31.1 (40%)	65	+2q22.2-q37.3 (25%)	Mosaic
78	Re-biopsy	HB 2-2	6	L	-13q31.1-q34	33	-13q31.1-q34	ABN
79	Discard	HB 2-3	6	G complex	+3q26.32-q29, +3p26.3-q26.31 (55%)	21	na	na
80#	Re-biopsy	HB 2-2	5	L	-7q31.1-q36.3 (80%), -7p22.3-q22.3 (40%)	50	NAD	Mosaic
81*	Re-biopsy	HB 2-3	6	L complex	-7q35-q36.3, +7p22.3-q35 (20%)	11	-7q35-q36.3, +7p22.3-q33 (50%)	ABN
82*	Re-biopsy	HB 2-2	6	L complex	-5q11.2-q35.3, -14q21.1-q32.33	130	-5q11.2-q35.3, -14q21.1-q32.33 (60%)	ABN
83	Re-biopsy	HB 2-3	5	G complex	+17q12-q21.32, +19p13.2-p11	12, 13	NAD	Mosaic
84	Discard	HB 2-2	6	G complex	+18p11.32-p11.21, +18q11.2-q23 (40%)	17	na	na
85	Discard	HB 1-2	5	L complex	-2q14.3-q37.3, -14q24.1-q32.33	108, 36	na	na

86	Discard	HB 2-3	6	L	-11q23.1-q25	35	na	na
87 <sup>#</sup>	Re-biopsy	FHB 2-2	6	L	-1q21.2-q44 (80%)	89	NAD	Mosaic
88*	Re-biopsy	HB 1-2	5	L complex	-5q23.2-q35.3, -5p15.33-p12 (50%)	47	-5q23.2-q35.3, +5q11.2-q23.1 (55%)	ABN
89	Re-biopsy	HB 2-2	5	G complex	+1p36.33-q31.2, +1q31.2-q44 (60%)	180	NAD	Mosaic
90	Discard	HB 2-2	6	G	+4q21.3-q35.2	104	na	na
91	Discard	HB 2-3	6	G	+14q21.3-q32.33	59	na	na
92	Discard	HB 2-2	5	L	-19q12.2-q13.43	41	na	na
93	Re-biopsy	HB 2-2	5	G	+6q21-q27	55	NAD	Mosaic
94	Re-biopsy	HB 1-1	5	G	+8q23.3-q24.3 (200%)	33	NAD	Mosaic
95 <sup>#</sup>	Re-biopsy	HB 1-1	5	G	+9q13-q34.3 (80%)	69	NAD	Mosaic
96	Re-biopsy	HB 2-2	5	L	-11q12.1-q25	79	-11q12.1-q25	ABN
97**#	Re-biopsy	HB 1-2	5	L complex	-4q13.3-q35.2 (90%), -4p16.3-q13.2 (40%)	121	-4q21.21-q35.2, -4p16.3-q21.21 (30%)	ABN
98	Discard	FHB 2-2	6	L	-1p36.33-p36.13	20	na	na
99	Re-biopsy	HB 2-2	5	G	+7p22.3-p21.3	11	NAD	Mosaic
100	Re-biopsy	HB 2-2	5	L	-2q23.1-q37.3	95	NAD	Mosaic
101	Re-biopsy	HB 2-2	5	G	+4p16.3-p16.1	11	NAD	Mosaic
102**#	Re-biopsy	HB 2-2	6	L complex	-4q22.3-q35.2 (90%), -4p16.3-q22.2 (65%)	94	-4q22.3-q35.2 (50%)	Mosaic
103*	Re-biopsy	HB 2-2	5	L complex	-11q13.3-q25, -11p15.5-q13.2 (20%)	66	-11q13.3-q25, -11p15.5-q13.2 (40%)	ABN
104*	Re-biopsy	HB 2-2	6	G	+8q21.3-q24.3	49	-8q21.3-q24.3, -8p22.3-q21.3 (70%)	ABN
105*	Re-biopsy	HB 2-2	5	L complex	-7p22.3-p21.3, -7p21.3-q36.3 (25%)	10	-7	ABN
106*	Re-biopsy	HB 2-3	6	G complex	+9p23-q34.3, +9p24.3-p23 (35%)	129	+9 (200%)	ABN
107**#	Re-biopsy	FHB 1-2	6	L	-20p13-p11.21 (85%)	25	+20p13-p11.21	ABN
108*	Re-biopsy	HB 2-2	5	G	+9p24.3-q12	57	+9	ABN
109*	Re-biopsy	HB 1-2	5	L	-1p36.33-p36.31	8	-1p36.33-p36.31 (50%)	Mosaic
110*	Re-biopsy	HB 2-3	5	L complex	-11q13.4-q25, -11p15.5-q13.4 (55%)	62	-11q13.4-q25 (70%)	ABN
111*	Re-biopsy	FHB 2-2	6	L	-5p15.33-p15.2, +p15.1-p13.1 (30%)	14	-5p15.33-p15.2	ABN

<sup>#</sup>: originally classified as ABN on the basis of an 80-95% shift, \*: the 2 TE samples exhibited discrepancies in the number, type, position, or magnitude of the SAs detected, <sup>a</sup> G: gain, L: loss, B: both (G + L), complex: additional mosaic shifts on same chromosome or same type of SA on a different chromosome, <sup>b</sup> shifts >100% indicate a gain of more than 1 copy (eg. 1800% = 18 extra copies of the segment observed), <sup>c</sup> the indicated size in Mb only refers to shifts ( $\geq 80\%$ ) classified as ABN, the sizes of mosaic shifts (<80%) are not given, <sup>d</sup> na: not applicable, NAD: no abnormality detected, <sup>e</sup> ABN: Abnormal.

**Supplementary table 7 Clinical re-biopsy outcomes of revised protocol**

	Simple gains (80-100%)	Other SA (80-95% shifts)	Total
Number thawed (% of total thawed)	44 (75.9%)	14 (24.1%)	58
Did not become suitable for re-biopsy (% of thawed)	4 (9.5%)	1 (9.1%)	5 (8.6%)
New result obtained (% of thawed)	40 (90.9%)	13 (92.9%)	53 (91.4%)
Confirmed ABN (% of new results)	11 (27.5%)	4 (30.8%)	15 (28.3%)
Reclassified as mosaic (% of new results)	29 (72.5%)	9 (69.2%)	38 (71.7%)
Reclassified mosaic, no SA ≥20% in re-biopsy <sup>a</sup> (% of reclassified)	24 (82.8%)	5 (55.6%)	29 (76.3%)
Reclassified mosaic, mosaicism in re-biopsy <sup>a</sup> (% of reclassified)	5 (17.2%)	4 (44.4%)	9 (23.7%)

<sup>a</sup> includes any mosaic abnormality (20-80%) involving the same chromosome as that of the original SA.